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FÜR PRÄNATALE
DIAGNOSTIK

Information on first trimester anatomy scan

You came to our office for a first trimester anatomy scan with nuchal translucency measurement.
We would like to inform you on the objectives and limitations of this exam.

OBJECTIVE OF THE EXAM

Most pregnancies are uneventful and end with the birth of a healthy child. Rarely problems occur.
With today's exam we would like to answer three important questions:

1. **Is the child's body built normally?**
2. **What is the risk for the baby having a trisomy (e. g. trisomy 21 = Down syndrome)**
3. **What is the risk for complications during the pregnancy like preeclampsia or intrauterine growth retardation?**

An early answer to these questions gives you enough time for decisions in case of a serious anomaly. In case of an increased risk for preeclampsia this can be reduced by a treatment which has to be started before 16 weeks of gestation.

1. **Is the child's body built normally?**

   Regardless of the age of the mother, 2 % of all newborns have a congenital malformation, for instance a congenital heart defect. By a detailed sonogram at the time of the first trimester screening it is possible to detect 50 % of all congenital malformations before 14 weeks of gestation. Many but not all of these malformations can be treated so that the parents have the opportunity to prepare themselves before the birth of the child.

2. **What is the risk for the baby having a trisomy (e. g. trisomy 21 = Down syndrome)?**

   We know that the risk for having a baby with a trisomy rises with the age of the mother. Trisomy means that of the 23 pairs of chromosomes, one is not a pair but a group of three. The most common trisomy is trisomy 21, which is also called Down syndrome.

AGE RELATED RISK FOR DOWN SYNDROME AT 12 WEEKS OF GESTATION

20 years	1 : 1068	36 years	1 : 196
25 years	1 : 946	38 years	1 : 117
30 years	1 : 626	40 years	1 : 68
32 years	1 : 461	42 years	1 : 38
34 years	1 : 312	44 years	1 : 21

When calculating the risk for trisomies we start out with the age-related risk. By ultrasound we examine several markers of the unborn child. The presence or absence of these markers lowers the risk in most cases, but rarely it also increases the risk. Important markers are the nuchal translucency and the fetal nasal bone:

Nuchal translucency

The nuchal translucency is a collection of fluid under the skin behind the fetal neck. It can be seen in every unborn child between 11 and 13 weeks of gestation. The enlargement of the nuchal translucency increases the risk for the presence of various fetal conditions e. g. fetal heart defects, trisomies and others. However, it is important to know that an enlarged nuchal translucency does not mean that there will be a problem. **The majority of all fetuses with an enlarged nuchal translucency will be born as healthy babies.**

Nasal bone

In most fetuses with Down syndrome the nasal bone is either absent or too small at this gestational age. If we observe this in the ultrasound exam the risk for the presence of a Down syndrome rises.

If every first-trimester-screening which results in an elevated risk for trisomies is followed by additional testing, the detection rate for Down syndrome is approximately 75 %. Depending on the results of the first-trimester-screening we will either recommend non-invasive-testing (NIPT), a maternal blood test, or invasive testing of either the placenta (chorionic villous sampling CVS) or the amniotic fluid (amniocentesis). It is your decision whether you want additional testing and which testing you opt for. We will counsel you accordingly and you will have as much time for the decision as you need.

Non-invasive prenatal testing (NIPT)

This blood test analyses the cellfree DNA in the maternal blood. This test can detect 99 % of all fetuses with Down syndrome and 98–99 % of all fetuses with trisomy 18 or 13. It is important to know, however, that this so called **non-invasive prenatal test** examines the placental DNA and not the fetal DNA. Typically they are identical but there are instances where the placental DNA and the fetal DNA are not identical. For this reason, false positive results may occur, especially in younger mothers. Furthermore this test cannot detect bodily malformations, which represent the majority of all fetal abnormalities. This test can be a useful addition to the sonographic exam but it cannot replace it. The **non-invasive prenatal test** cannot detect abnormalities in other chromosomes besides 21, 18, 13, X and Y. Therefore its performance is lower than that of chorionic villous sampling or amniocentesis. An abnormal result in a **non-invasive prenatal test** has to be confirmed by amniocentesis. The test is covered by public health insurance. After the ultrasound exam the doctor will discuss with you whether this test is advisable for you.

Summary

A fetal trisomy can only be definitely excluded by counting the chromosomes of the fetal cells under the microscope. Fetal cells can be sampled from the placenta (after 11 completed weeks) or the amniotic fluid (after 16 completed weeks). The risk calculation described above enables all women with a low risk for trisomies to avoid invasive testing. This applies to 95–97 % of all pregnancies.

3. What is the risk for complications during the pregnancy like preeclampsia or intrauterine growth retardation?

Preeclampsia is a complication occurring in the second half of the pregnancy. Typical symptoms are high blood pressure and excretion of protein in the urine. There is often a placental insufficiency so that the child has to be delivered earlier, sometimes very prematurely. Preeclampsia occurs in 3 out of 100 pregnancies and in 3 out of 1000 it occurs before 34 weeks of gestation.

The risk for developing preeclampsia can be calculated from the blood flow in the maternal arteries leading to the uterus, the placental growth factor in the maternal blood (PlGF) and the maternal history. In case of an increased risk a treatment with low-dose aspirin can prevent 60 – 80 % of all cases of preeclampsia. Testing for PlGF is not covered by the insurance and costs approximately 60 Euros. After the ultrasound the doctor will tell you, whether testing for PlGF is recommended in your situation.

PLEASE BE AWARE OF THE FOLLOWING:

Today's exam cannot exclude all fetal malformations, fetal abnormalities or fetal chromosomal abnormalities. A normal first-trimester screening cannot guarantee a healthy child. For chromosomal abnormalities we can only give you a risk calculation and not a definite diagnosis. A definite diagnosis for chromosomal abnormalities is only possible by invasive testing.

Most exams do not show any abnormalities. This leads to reassurance and to an undisturbed course of the pregnancy.

In case of the detection of abnormalities, this can cause substantial anxiety and conflicts. We will inform you as detailed as possible and arrange consultations with other physicians like geneticists, pediatricians or pediatric surgeons. We also recommend psychosocial counselling in this situation and can arrange this if you wish.

GENETIC COUNSELLING

Before the exam we will ask you whether there are persons with congenital problems or malformations in your family history. This allows us to assess your risk for having children with hereditary health problems. If we happen to find any significant history we may refer you to a geneticist for genetic counselling.

Of course you have the choice as to whether to answer the questions concerning the family history.

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QUESTIONNAIRE

Name _____ **First name** _____

born _____

Patient Number _____

Please take some time to answer the following questions:

Address _____
Mobile Number _____
Phone Number _____
E-mail _____
Health insurance _____
Gynecologist _____
Profession _____
Reason for referral _____

General history

How tall are you? _____ cm

What was your weight before the pregnancy? _____ kg

What is your present weight? _____ kg

Do you smoke? YES ☐ NO ☐ if yes, how many: _____

Do you drink alcohol? YES ☐ NO ☐

Present pregnancy

First day of last menstrual period: _____ Due date: _____

Did you get pregnant with the help of fertility treatment? YES ☐ NO ☐

If yes, which one (e.g. IVF/ICSI/insemination)? _____

If yes, did you use your own egg cells? YES ☐ NO ☐

Do or did you take any medication during the pregnancy? YES ☐ NO ☐

If yes, please specify. _____

Have you had a non-invasive prenatal test done? (NIPT, e.g. Praenatest, Harmony)? YES ☐ NO ☐ if yes, which test: _____

result _____

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Previous pregnancies

Have you had any **miscarriages?**	YES ☐ NO ☐	if yes, how many:	_____
Have you had any miscarriages after 16 weeks of gestation?	YES ☐ NO ☐		
Have you had any **terminations of pregnancy?**	YES ☐ NO ☐	if yes, how many:	_____
Was a health problem of the unborn child the reason for one of the terminations?	YES ☐ NO ☐		
Was one of the terminations performed after 16 weeks of gestation?	YES ☐ NO ☐		
Have you had an **extrauterine pregnancy?**	YES ☐ NO ☐	if yes, how many:	_____

Questions concerning the risk calculation for pregnancy complications

Were you pregnant previously?	YES ☐ NO ☐	if yes, how often:	_____
Did you give birth?	YES ☐ NO ☐	if yes, how often:	_____
Was there a **chromosomal abnormality** in one of your previous pregnancies?	YES ☐ NO ☐	if yes, which abnormality:	_____
Are all children healthy?	YES ☐ NO ☐		

If it applies:

Vaginal deliveries between 16 and 30 weeks _____

Vaginal deliveries between 30 and 36 weeks _____

Deliveries after 37 completed weeks _____

DOB of your youngest child _____

Do you suffer from a **chronic health condition** (e.g. diabetes, hypertension, malfunction of the thyroid)? YES ☐ NO ☐

If yes, please specify. _____

Do you suffer from a **rheumatic disease** (e.g. lupus erythematodes, antiphospholipid syndrome (APS))? YES ☐ NO ☐

If yes, please specify. _____

Did you ever have **preeclampsia?** YES ☐ NO ☐

Did anyone in your family have preeclampsia? YES ☐ NO ☐

Was one of your children too small at birth? YES ☐ NO ☐

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Please give us additional information, if you have children:

Year of birth	Vaginal delivery (n) or c-section (K)	weight	Delivery before 37 weeks? If yes, which week?	Additional information you would like to provide?

Family history

In your family, are there persons with congenital problems, malformations or handicaps? YES ☐ NO ☐

If yes, please name the problem and the relationship: _____

Did anybody in your or your partners family have two or more miscarriages? YES ☐ NO ☐

The child's father

In the family of the child's father, are there persons with congenital problems, malformations or handicaps? YES ☐ NO ☐

If yes, please name the problem and the relationship: _____

Are you related to the child's father (e.g. cousin)? YES ☐ NO ☐

If yes, what is the relationship? _____

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Consent to the exam according to the german law for genetic diagnosis (Gendiagnostikgesetz GenDG)

I hereby confirm that I have read and understood the information on the first trimester screening	YES ☐	NO ☐
I want to be informed on markers that indicate chromosomal abnormalities	YES ☐	NO ☐
I would give birth to a child with Down syndrome	YES ☐	NO ☐
I wish a counselling by a geneticist if the sonogram is abnormal	YES ☐	NO ☐
I wish a counselling by a geneticist before the sonogram (requires a new appointment)	YES ☐	NO ☐
I consent that the results of the exam are given to		
my partner	YES ☐	NO ☐
my doctor	YES ☐	NO ☐

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Do you have questions or is there anything you would like to add?

Additional remarks by the doctor:

I have been informed on the upcoming exam verbally as well as in writing.

All my questions concerning the exam have been discussed and have been answered so that I could understand.
I feel well informed, do not have any additional questions and consent to the exam.

☐ I need more time to think about the exam
☐ I do not need more time to think about the exam

Berlin, _____ signature of the patient

Berlin, _____ signature of the doctor